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SEQUENCE LISTING

<110> Weissenbach, Jean
Hazan, Jamilé

<120> CLONING, EXPRESSION AND CHARACTERIZATION OF THE SPG4 GENE RESPONSIBLE FOR THE MOST COMMON FORM OF AUTOSOMAL DOMINANT SPASTIC PARAPLEGIA

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Replacement Sheet

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Replacement Sheet

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<400> 55
gcacttaacc aggctgtatg 20

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<400> 57
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gcaataattc accacacttg 20

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ggtagttctt gtttctgctc 20

<210> 61
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<220>
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<400> 61
caagtgtggt gaattattgc 20

<210> 62
<211> 19
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<220>
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<220>
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<220>
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agcctctgga gatagtatgc 20

<210> 65
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<220>
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ctagaacagg ggtcacagtc 20

<210> 66
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<220>
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<400> 66
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<210> 67
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<220>
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<400> 67
gcagtatgca agaaattgaa c 21

<210> 68
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<220>
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<400> 68
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<220>
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<400> 70
gtgtagcaga tcaacatag 19

<210> 71
<211> 20
<212> DNA
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<220>
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<400> 71

catcttcaag tttggtgcac 20

<210> 72
<211> 1689
<212> DNA
<213> mouse

<220>
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gtatcgaaga actggaaaaa ggaatcgctg ttatagttac gggccaaggt gaacagtatg 180
aaagagctag acgtcttcaa gccaaaatga tgactaattt agttatggcc aaggaccgtt 240
tacaacttct agagaagctg caaccagttt tgcaattttc caagtcacag acggacgtct 300
ataacgagag tactaacctg acatgccgca atggacatct ccagtcagaa agtggagcag 360
ttccgaagag gaaagacccc ttaacacatg ctagtaattc attgcctcga tcaaaaactg 420
tcctgaaaag tggctccgca gggctctccg gtcaccacag ggcgcctagt tgcagtggtt 480
tgtccatggt ttctggagca agaccgggac ctggtcctgc agctaccaca cataagggta 540
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aagacttgaa aaattttagg aatgtggaca gcaatcttgc taaccttata atgaatgaaa 660
ttgttgacaa tgggacagct gttaagtttg atgacatagc cgggcaggag ctggcaaagc 720
aagcgctgca ggagattgtc atccttcctt ctctgcggcc tgagttgttc acagggctca 780
gagctcctgc tagaggcttg ttactcttcg gtccgccagg aaacggaaaa acaatgctgg 840
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taaaacgcag tgtgagtcct cagaccttag aagcatacat acgctggaac aaggattttg 1500
gagacaccac tgtttaaagg aatggatgcc tctgtgagcc catagaacat cgcacttcac 1560
aggaaacaag agctttggct acaggaaccc agacttcgtt tacaggacgt tttagagttt 1620
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tatgaatgg 1689

<210> 73
<211> 504
<212> PRT
<213> mouse

<220>
<223> Incomplete murine spastin.

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Ile Ala Leu Arg Ile Asp Glu Glu Glu Lys Ala Gly Gln Lys Glu Gln
20 25 30

Replacement Sheet

Ala Val Glu Trp Tyr Lys Lys Gly Ile Glu Glu Leu Glu Lys Gly Ile
35 40 45

Ala Val Ile Val Thr Gly Gln Gly Glu Gln Tyr Glu Arg Ala Arg Arg
50 55 60

Leu Gln Ala Lys Met Met Thr Asn Leu Val Met Ala Lys Asp Arg Leu
65 70 75 80

Gln Leu Leu Glu Lys Leu Gln Pro Val Leu Gln Phe Ser Lys Ser Gln
85 90 95

Thr Asp Val Tyr Asn Glu Ser Thr Asn Leu Thr Cys Arg Asn Gly His
100 105 110

Leu Gln Ser Glu Ser Gly Ala Val Pro Lys Arg Lys Asp Pro Leu Thr
115 120 125

His Ala Ser Asn Ser Leu Pro Arg Ser Lys Thr Val Leu Lys Ser Gly
130 135 140

Ser Ala Gly Leu Ser Gly His His Arg Ala Pro Ser Cys Ser Gly Leu
145 150 155 160

Ser Met Val Ser Gly Ala Arg Pro Gly Pro Gly Pro Ala Ala Thr Thr
165 170 175

His Lys Gly Thr Pro Lys Pro Asn Arg Thr Asn Lys Pro Ser Thr Pro
180 185 190

Thr Thr Ala Val Arg Lys Lys Lys Asp Leu Lys Asn Phe Arg Asn Val
195 200 205

Asp Ser Asn Leu Ala Asn Leu Ile Met Asn Glu Ile Val Asp Asn Gly
210 215 220

Thr Ala Val Lys Phe Asp Asp Ile Ala Gly Gln Glu Leu Ala Lys Gln
225 230 235 240

Ala Leu Gln Glu Ile Val Ile Leu Pro Ser Leu Arg Pro Glu Leu Phe
245 250 255

Thr Gly Leu Arg Ala Pro Ala Arg Gly Leu Leu Leu Phe Gly Pro Pro
260 265 270

Gly Asn Gly Lys Thr Met Leu Ala Lys Ala Val Ala Ala Glu Ser Asn
275 280 285

Ala Thr Phe Phe Asn Ile Ser Ala Ala Ser Leu Thr Ser Lys Tyr Val
290 295 300

Gly Glu Gly Glu Lys Leu Val Arg Ala Leu Phe Ala Val Ala Arg Glu
305 310 315 320

Leu Gln Pro Ser Ile Ile Phe Ile Asp Glu Val Asp Ser Leu Leu Cys
325 330 335

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Glu Arg Arg Glu Gly Glu His Asp Ala Ser Arg Arg Leu Lys Thr Glu
340 345 350

Phe Leu Ile Glu Phe Asp Gly Val Gln Ser Ala Gly Asp Asp Arg Val
355 360 365

Leu Val Met Gly Ala Thr Asn Arg Pro Gln Glu Leu Asp Glu Ala Val
370 375 380

Leu Arg Arg Phe Ile Lys Arg Val Tyr Val Ser Leu Pro Asn Glu Glu
385 390 395 400

Thr Arg Leu Leu Leu Leu Lys Asn Leu Leu Cys Lys Gln Gly Ser Pro
405 410 415

Leu Thr Gln Lys Glu Leu Ala Gln Leu Ala Arg Met Thr Asp Gly Tyr
420 425 430

Ser Gly Ser Asp Leu Thr Ala Leu Ala Lys Asp Ala Ala Leu Gly Pro
435 440 445

Ile Arg Glu Leu Lys Pro Glu Gln Val Lys Asn Met Ser Ala Ser Glu
450 455 460

Met Arg Asn Ile Arg Leu Ser Asp Phe Thr Glu Ser Leu Lys Lys Ile
465 470 475 480

Lys Arg Ser Val Ser Pro Gln Thr Leu Glu Ala Tyr Ile Arg Trp Asn
485 490 495

Lys Asp Phe Gly Asp Thr Thr Val
500

<210> 74
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 74
atttttttatt ttaaagcagg acag 24

<210> 75
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 75
aatttttttc tttcaggtga acag 24

<210> 76
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 76
cttctctgtt gcatagagaa gatg 24

<210> 77
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 77
acttttttcct tgtcagaaag tgga 24

<210> 78
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 78
ttttgtatcc tttaagggta ctcc 24

<210> 79
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 79
aggtcttgtt tcttagtgga acag 24

<210> 80
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 80
agtatatatt ttttagttgt tcac 24

<210> 81
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 81
cttgtgattt ttaaaggcta aagc 24

<210> 82
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 82
taatgctttg ttttaggtgg gaga 24

<210> 83
<211> 24
<212> DNA
<213> Homo sapiens

<220>
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<400> 83
cttgtatttc ctctagatga agtt 24

<210> 84
<211> 24
<212> DNA
<213> Homo sapiens

<220>
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<400> 84
gatttttttgc ttgtaggtac agtc 24

<210> 85
<211> 24

<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 85
ggatttttt ttttaggcgt ttca 24

<210> 86
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 86
ttttaatatt tttcagacaa gact 24

<210> 87
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 87
tccttccctt cctcagaatg actg 24

<210> 88
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 88
cttttatgtt ttacagaact aaaa 24

<210> 89
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice acceptor site.

<400> 89
ctttttaaaa atctagatga gaaa 24

<210> 90
<211> 24
<212> DNA
<213> Homo sapiens

<220>
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<400> 90
tgagaaaggt aactaggggg ctgg 24

<210> 91
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

<400> 91
aggacaaggt aagattgtat ttgt 24

<210> 92
<211> 24
<212> DNA
<213> Homo sapiens

<220>
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<400> 92
acttctaggt atcaattaat gtat 24

<210> 93
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<220>
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<400> 93
ccagtcaggt gggtttaggt taac 24

<210> 94
<211> 24
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<220>
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<400> 94
ctcataaggt attctgggac agta 24

<210> 95
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

<400> 95
gtggacaagt aagttttgcc atct 24

<210> 96
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<213> Homo sapiens

<220>
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<400> 96
ggcctgaggt aagaacttta tatt 24

<210> 97
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

<400> 97
caatgctggt aagggttctc ttca 24

<210> 98
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

<400> 98
caaaatacgt gagtgctctg tttc 24

<210> 99
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<212> DNA
<213> Homo sapiens

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<400> 99
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<210> 100
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

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<210> 101
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<212> DNA
<213> Homo sapiens

<220>
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gttctcaggt agggagattt atat 24

<210> 102
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<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

<400> 102
atgaggaggt atgtatctgt gttt 24

<210> 103
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

<400> 103
cttgctaggt gagtaatttg gatt 24

<210> 104
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

<400> 104
tatccgaggt aggtatacaa gagc 24

<210> 105
<211> 24
<212> DNA
<213> Homo sapiens

<220>
<223> SPG4 gene splice donor site.

<400> 105
ccagtgaggt atagtatttt acaa 24

<210> 106
<211> 2152
<212> DNA
<213> Mus musculus

<220>
<223> Complete Spg4 DNA

<400> 106
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acttgaagag gaacaagaag acagacctaa ataaaatatg caatatgaat gg 2152

<210> 107
<211> 614
<212> PRT
<213> Mus musculus

<220>
<223> Complete murine spastin

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20 25 30

Ala Ala Gly Pro Ala Pro Ala Ala Gly Ser Pro Pro Lys Arg Asn Leu
35 40 45

Ser Ser Phe Ser Ser Pro Leu Val Val Gly Phe Ala Leu Leu Arg Leu
50 55 60

Leu Ala Cys His Leu Gly Leu Leu Phe Ala Trp Leu Cys Gln Arg Phe
65 70 75 80

Ser Arg Ala Leu Met Ala Ala Lys Arg Ser Ser Gly Thr Ala Pro Ala
85 90 95

Pro Ala Ser Pro Ser Pro Pro Ala Pro Gly Pro Gly Gly Glu Ala Glu
100 105 110

Ser Val Arg Val Phe His Lys Gln Ala Phe Glu Tyr Ile Ser Ile Ala
115 120 125

Leu Arg Ile Asp Glu Glu Glu Lys Ala Gly Gln Lys Glu Gln Ala Val
130 135 140

Glu Trp Tyr Lys Lys Gly Ile Glu Glu Leu Glu Lys Gly Ile Ala Val
145 150 155 160

Ile Val Thr Gly Gln Gly Glu Gln Tyr Glu Arg Ala Arg Arg Leu Gln
165 170 175

Ala Lys Met Met Thr Asn Leu Val Met Ala Lys Asp Arg Leu Gln Leu
180 185 190

Leu Glu Lys Leu Gln Pro Val Leu Gln Phe Ser Lys Ser Gln Thr Asp

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        195                 200                 205

Val Tyr Asn Glu Ser Thr Asn Leu Thr Cys Arg Asn Gly His Leu Gln
    210                 215                 220

Ser Glu Ser Gly Ala Val Pro Lys Arg Lys Asp Pro Leu Thr His Ala
225                 230                 235                 240

Ser Asn Ser Leu Pro Arg Ser Lys Thr Val Leu Lys Ser Gly Ser Ala
                245                 250                 255

Gly Leu Ser Gly His His Arg Ala Pro Ser Cys Ser Gly Leu Ser Met
            260                 265                 270

Val Ser Gly Ala Arg Pro Gly Pro Gly Pro Ala Ala Thr Thr His Lys
        275                 280                 285

Gly Thr Pro Lys Pro Asn Arg Thr Asn Lys Pro Ser Thr Pro Thr Thr
    290                 295                 300

Ala Val Arg Lys Lys Lys Asp Leu Lys Asn Phe Arg Asn Val Asp Ser
305                 310                 315                 320

Asn Leu Ala Asn Leu Ile Met Asn Glu Ile Val Asp Asn Gly Thr Ala
                325                 330                 335

Val Lys Phe Asp Asp Ile Ala Gly Gln Glu Leu Ala Lys Gln Ala Leu
            340                 345                 350

Gln Glu Ile Val Ile Leu Pro Ser Leu Arg Pro Glu Leu Phe Thr Gly
        355                 360                 365

Leu Arg Ala Pro Ala Arg Gly Leu Leu Leu Phe Gly Pro Pro Gly Asn
    370                 375                 380

Gly Lys Thr Met Leu Ala Lys Ala Val Ala Ala Glu Ser Asn Ala Thr
385                 390                 395                 400

Phe Phe Asn Ile Ser Ala Ala Ser Leu Thr Ser Lys Tyr Val Gly Glu
                405                 410                 415

Gly Glu Lys Leu Val Arg Ala Leu Phe Ala Val Ala Arg Glu Leu Gln
            420                 425                 430

Pro Ser Ile Ile Phe Ile Asp Glu Val Asp Ser Leu Leu Cys Glu Arg
        435                 440                 445

Arg Glu Gly Glu His Asp Ala Ser Arg Arg Leu Lys Thr Glu Phe Leu
    450                 455                 460

Ile Glu Phe Asp Gly Val Gln Ser Ala Gly Asp Asp Arg Val Leu Val
465                 470                 475                 480

Met Gly Ala Thr Asn Arg Pro Gln Glu Leu Asp Glu Ala Val Leu Arg
                485                 490                 495

Arg Phe Ile Lys Arg Val Tyr Val Ser Leu Pro Asn Glu Glu Thr Arg
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Leu Leu Leu Leu Lys Asn Leu Leu Cys Lys Gln Gly Ser Pro Leu Thr
        515                 520                 525

Gln Lys Glu Leu Ala Gln Leu Ala Arg Met Thr Asp Gly Tyr Ser Gly
    530                 535                 540

Ser Asp Leu Thr Ala Leu Ala Lys Asp Ala Ala Leu Gly Pro Ile Arg
545                 550                 555                 560

Glu Leu Lys Pro Glu Gln Val Lys Asn Met Ser Ala Ser Glu Met Arg
                565                 570                 575

Asn Ile Arg Leu Ser Asp Phe Thr Glu Ser Leu Lys Lys Ile Lys Arg
            580                 585                 590

Ser Val Ser Pro Gln Thr Leu Glu Ala Tyr Ile Arg Trp Asn Lys Asp
        595                 600                 605

Phe Gly Asp Thr Thr Val
    610
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